ABC NEWS

A Tale of Two Sisters: Treatment for Rare Genetic Disease Offers New Hope

Pompe Disease Affects About 10,000 People Worldwide

June 14, 2006 — - Sisters Megan and Kelsey Assink were born with a rare genetic disease that kills most children during infancy.

But there was one key difference between the two girls -- time.

Megan and Kelsey were born eight years apart. The medical advancements made in that time gave one sister a chance to live that the other did not have.

"You just keep on holding on," said Deb Assink, the girls' mother. "Just keep hoping because without that, then, you know, there's nothing there."

Deb and Greg Assink, the girls' father, welcomed Kelsey, their first child, into the world in 1994.

"We noticed that she was behind, that she was delayed," Deb Assink said.

Doctors found that Kelsey had an enlarged heart, and more tests led to the diagnosis of a rare and fatal genetic disorder called Pompe disease.

The condition, which affects less than 10,000 people worldwide, is caused by a lack of an enzyme that is essential for normal muscle development, including the heart.

"Sugars that usually are used for energy build up inside the cells and cause weakness in the muscles," said Barry Byrne, professor of molecular genetics and gene therapy of cardiovascular disease at the University of Florida in Gainesville.

Medical Timing

In 1994, there was no treatment for Kelsey or other children like her.

Most with the infantile form of the disease died from respiratory or cardiac complications before 2 years of age.

Those who were diagnosed with the juvenile form of the disease -- usually in their second year of life -- had a life expectancy into their 20s. Those with the adult form of Pompe disease often lived long lives, although not as long as the average life expectancy.

"For these children, all that could be done was to try to maximize their quality of life until they died," said David Freyer, assistant professor of pediatric hematology-oncology at DeVos Children's Hospital in

Grand Rapids, Mich.

The Assinks had to just wait and see how Kelsey coped with her disease.

"It was just waiting," Deb Assink said. "We just had to wait."

Not a Death Sentence

Kelsey died at age 9. During her brief life, she lived at home on a ventilator, her muscles slowly deteriorating. She never walked, but lived well beyond doctors' expectations.

The Assinks had three more children -- Tyler, Hope and Megan.

Tyler and Hope were healthy, but Megan was diagnosed with Pompe disease when she was 6 months old. Kelsey died months after Megan's diagnosis.

"That Sunday we disconnected her from her trach and that was very difficult," Deb Assink said. "It was a beautiful Sunday. It was a rainy and gray [day], and then when we did that the sun came up."

At Kelsey's funeral, the Assinks learned from a friend that baby Megan was not necessarily sentenced to her sister's fate. Megan was eligible for an experimental treatment called Myozyme, an enzyme replacement therapy that was the first of its kind.

At 18 months old, Megan was one of the first patients to receive the enzyme replacement in clinical trials. As the therapy continued and time passed, Megan's muscles began to get stronger.

"She went from not walking, not being able to put weight on her legs to crawling all over and pulling herself up onto her legs," Deb Assink said.

Doctors were most impressed by the improvements in Megan's heart, which no longer showed evidence of disease.

A Happy Ending

Today, Megan is a happy 4 year old.

Every two weeks, for an entire day, Megan receives a transfusion of the enzyme her body doesn't make.

Her progress has been remarkable. She walks -- a little unsteadily at times -- but can keep up with her sister, Hope. Speaking is difficult, but she is working with a therapist to help strengthen her weak facial muscles.

On April 28, 2006, the Food and Drug Administration approved the use of Myozyme, the first treatment ever approved for Pompe disease.

While the treatment offers hope, it also comes at a high cost. Without insurance or support of the company that makes the treatment, the cost is more than $100,000 a year.

Because Megan is one of the first people to receive this treatment, doctors don't know whether her progression will continue. It is also unclear whether she will require fewer treatments as time goes on.

Megan's parents are just happy she has a chance that Kelsey never had.